WES CRANIOFACIAL ANOMALIES DG 3.2

Gene	Median coverage	% covered >10x	% covered >20x	OMIM disease ID
ACP4	95.6	98.4	91.4	617297
ADAMTSL4	138.1	99.8	98.4	225100
ALX1	131.8	99.6	95.2	613456
ALX3	124.2	80.2	72.8	136760
ALX4	134.4	100.0	99.9	168500
AMBN	164.9	99.6	97.7	616270
AMELX	86.8	99.4	94.0	301200
AMER1	96.7	99.6	96.6	300373
AMTN	116.5	99.9	99.1	617607
ANKRD11	118.2	97.0	94.0	148050
ARHGAP29	144.0	98.9	97.6	-
AXIN2	125.3	100.0	99.8	608615
BCOR	99.9	99.2	95.8	300166
BMP2	167.5	100.0	100.0	-
BMP4	175.3	100.0	100.0	607932
CCBE1	73.7	99.9	98.8	No OMIM phenotype
CDC45	144.4	99.8	98.5	617063
CDON	118.7	99.9	98.6	614226
CDSN	123.1	100.0	99.8	602593
CHD7	137.1	100.0	99.2	214800
COL11A1	100.9	96.0	92.7	154780;604841
COL11A2	126.7	100.0	99.6	215150;277610
COL2A1	122.0	100.0	99.8	108300
COL9A1	129.1	99.9	98.6	614135
COL9A2	105.0	99.9	98.9	614284;600204

COL9A3	106.6	98.9	95.5	120270
COLEC11	169.1	100.0	100.0	No OMIM phenotype
CTSK	90.8	100.0	99.2	265800
CYP26B1	165.6	100.0	100.0	No OMIM phenotype
DHODH	106.1	100.0	99.9	263750
DISP1	160.1	99.9	99.9	-
DLX3	130.7	99.8	97.6	104510
DLX4	253.1	100.0	100.0	616788
DSPP	64.8	97.8	93.5	125490
EDA	100.0	98.3	89.7	305100;313500
EDAR	140.1	100.0	99.7	129490;224900
EDARADD	87.2	99.7	98.9	129490;224900
EDN1	135.4	100.0	99.4	615706
EDNRA	166.2	99.8	99.8	616367
EFNA4	149.9	100.0	100.0	601380
EFNB1	136.4	100.0	100.0	304110
EFTUD2	111.1	100.0	99.2	610536
EIF4A3	87.6	100.0	99.2	268305
ENAM	130.8	100.0	100.0	104500
ERF	138.1	100.0	98.9	600775
ESCO2	114.4	98.5	94.6	268300
EYA1	120.9	99.9	99.5	113650
EZH2	144.1	99.7	98.0	277590
FAM20A	103.8	99.6	94.4	204690
FAM83H	84.2	85.5	83.1	130900
FGD1	83.8	97.2	91.2	305400
FGF10	137.9	100.0	99.5	149730
FGF3	125.9	100.0	98.7	610706
FGF8	123.7	97.1	87.2	612702
FGF9	184.7	100.0	100.0	612961

FGFR1	128.6	100.0	99.3	123150
FGFR2	113.3	97.6	97.0	123500
FGFR3	124.7	99.8	98.0	101400
FLNA	143.7	100.0	99.9	No OMIM phenotype
FLNB	130.4	99.4	98.7	No OMIM phenotype
FOXC1	56.3	98.7	93.7	602482;601631
FOXE1	42.9	97.9	82.2	241850
GDF3	114.9	100.0	100.0	613702
GDF6	113.8	100.0	100.0	118100
GJA1	151.1	100.0	100.0	121014
GJB6	121.9	100.0	100.0	129500
GLI2	163.8	99.8	98.6	610829
GLI3	131.0	98.5	97.7	175700
GNAI3	102.5	98.4	93.2	602483
GNPTAB	148.1	99.9	99.7	No OMIM phenotype
GPR68	141.1	99.6	97.1	617217
GRHL3	151.8	100.0	99.9	606713
GSC	104.0	98.9	93.0	602471
HOXA2	76.2	99.9	99.3	612290
HUWE1	80.5	98.6	93.2	300706
HYAL2	175.2	100.0	99.9	-
IFT122	131.2	99.9	99.2	218330
IFT43	116.4	100.0	100.0	614099
IFT88	91.9	99.3	96.9	-
IKBKG	55.8	84.6	75.2	300291;300301
IL11RA	146.0	100.0	99.6	614188
IL6ST	97.3	94.9	89.4	No OMIM phenotype
IMPAD1	147.0	100.0	99.9	No OMIM phenotype
INTU	119.2	99.9	98.6	617926
IRF6	85.2	99.4	93.0	119300

ITGB6	133.8	97.4	95.7	616221
KAT6B	148.0	99.4	98.0	606170;603736
KDF1	104.0	99.8	98.0	617337
KDM1A	123.7	97.7	95.1	616728
KDM6A	98.7	94.2	85.9	300867
KLK4	191.1	100.0	100.0	204700
KMT2D	133.9	99.9	99.0	147920
KREMEN1	139.8	99.3	96.1	609898
LAMB3	120.5	99.9	98.8	104530
LRP2	147.6	100.0	99.8	222448
LRP6	137.0	100.0	99.2	616724
LTBP3	134.5	99.8	98.6	613097
MASP1	141.9	100.0	99.6	257920
MED12	88.0	99.3	94.1	300895;305450;309520
MEGF8	141.9	100.0	99.2	614976
MEIS2	122.2	100.0	99.7	600987
MEOX1	100.8	100.0	98.0	214300
MID1	137.3	99.6	97.7	300000
MITF	135.0	100.0	99.9	193510
MMP20	103.8	99.9	99.3	612529
MN1	119.7	100.0	99.7	No OMIM phenotype
MSX1	120.3	97.7	92.6	189500
MSX2	83.9	100.0	98.0	168500
NAA10	110.0	99.8	97.9	300013
NECTIN1	131.2	100.0	99.7	225060
NFKBIA	123.5	92.4	83.0	612132
NIPBL	119.2	98.4	96.3	122470
NOG	189.0	100.0	100.0	186500
NSD1	137.5	100.0	99.8	117550
ODAPH	242.0	100.0	100.0	614832

OFD1	50.3	87.1	71.3	311200
OTX2	124.0	100.0	99.0	610125
P4HB	116.1	94.6	94.0	No OMIM phenotype
PAX3	97.0	100.0	99.8	193500
PAX6	117.9	100.0	99.9	602482
PAX7	142.9	100.0	100.0	268220
PAX9	221.0	99.7	99.6	604625
PGM1	124.5	94.2	94.1	614921
PITX2	147.2	99.8	97.2	180500
PLCB4	99.9	99.8	98.7	614669
POLR1C	83.8	89.6	84.8	248390
POLR1D	172.9	91.6	91.6	613717
POR	193.1	99.5	98.0	No OMIM phenotype
PORCN	110.8	100.0	99.1	305600
PTCH1	110.2	99.3	96.6	109400
PTH1R	104.0	99.6	95.9	125350
RAB23	114.7	99.7	99.7	201000
RAD21	89.2	99.2	95.9	614701
RBM10	119.3	99.8	97.3	311900
RECQL4	158.3	99.9	98.6	603780
RIPK4	151.7	100.0	99.9	263650
RUNX2	105.6	72.2	72.2	119600
SALL1	117.9	99.7	97.5	107480
SALL4	137.8	99.1	96.4	607323
SATB2	102.1	99.5	96.5	612313
SCARF2	76.3	97.4	88.9	No OMIM phenotype
SEC24D	136.6	99.9	99.3	No OMIM phenotype
SEMA3E	137.7	99.1	98.9	214800
SF3B4	59.4	99.8	94.1	154400
SH3BP2	153.6	91.4	91.3	118400

SHH	125.7	100.0	100.0	147250;611638
SIX1	122.0	100.0	99.7	608389
SIX3	164.7	99.3	96.9	157170
SKI	110.2	99.7	97.1	182212
SLC24A4	114.6	100.0	99.5	615887
SLC26A2	199.3	100.0	100.0	256050
SMAD6	177.9	90.7	79.3	617439
SMC1A	89.7	99.6	97.1	300590
SMC3	79.3	94.5	89.0	610759
SMO	134.2	98.9	94.7	601707
SMOC2	86.5	76.7	74.9	125400
SNAI2	105.9	99.9	98.0	608890
SOX10	69.2	99.9	97.2	613266
SOX6	102.8	99.9	98.9	607257
SOX9	171.5	100.0	99.9	114290
SPECC1L	118.1	96.0	95.0	600251
SUMO1	19.5	60.5	45.3	613705
TBX1	91.3	87.4	77.6	192430
TBX22	116.2	98.4	93.8	303400
TCF12	136.8	99.9	99.7	615314
TCOF1	120.8	99.7	98.7	154500
TFAP2A	101.7	98.1	92.1	113620
TGFBR1	156.6	93.6	93.6	609192
TGFBR2	159.9	100.0	99.9	610168
TGIF1	133.3	100.0	100.0	142946
TLK2	93.9	98.5	93.2	No OMIM phenotype
TP63	170.6	100.0	100.0	604292
TRAF6	78.2	96.2	86.1	602355
TSHZ1	152.9	98.8	98.8	607842
TSPEAR	147.0	100.0	99.7	618180

TWIST1	102.3	100.0	99.4	101400
UBB	51.2	100.0	99.5	119540
VAX1	97.1	98.9	93.9	614402
WDR19	128.4	99.8	98.6	614378
WDR35	147.5	99.6	98.4	613610
WDR72	130.0	96.5	95.6	613211
WNT10A	131.1	100.0	98.9	224750;257980
WNT10B	145.8	100.0	99.6	617073
ZEB2	137.0	99.7	98.5	235730
ZIC1	240.6	100.0	100.0	616602
ZIC2	104.5	100.0	99.3	609637

Gene symbols used follow HGCN guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan 43(Database issue):D1079-85.
TWIST is the default chemistry for all WES samples. Agilent V5 was the default chemistry until Q3 2021.
Median Coverage describes the average number of reads seen across 50 exomes.
Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x.
Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x.
Genes with no value for coverage are non protein coding genes.
Non protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions: September 1st, 2021.